American Journal of Medical Genetics 62:318 (1996)

Letter to the Editor

Opitz GBBB Syndrome and the 22q11.2 Deletion

To the Editor:

Recently, McDonald-McGinn et al. [1995] reported the presence of a deletion 22q11.2 in a family with autosomal dominant inheritance and in a sporadic case with the Opitz GBBB syndrome.The presence of a vascular ring in these patients prompted them to look for this deletion, since this anomaly may be associated with the 22q11.2 deletion [Zackai et al., 1995]. They reviewed the Opitz GBBB syndrome and the 22q11.2 microdeletion syndrome finding considerable overlap of manifestations. They proposed that, in some patients, the Opitz GBBB syndrome may be due to a 22q11.2 deletion.

We recently (12/30/95) examined a newborn boy referred because of MCA. The cardinal findings in this patient (hypertelorism, hypospadias with descended testicles, characteristic nose and truncus arteriosus type I) were suggestive of the Opitz GBBB syndrome and of the velocardiofacial syndrome. The chromosomes were apparently normal (46,XY), but the FISH study showed a 22q11.2 deletion. The patient developed hypocalcemia with very low level of PTH and heart failure requiring surgery. His immunological status was normal except that CD4 cells were mildly low and natural killer cells were increased in number. The family history was non-contributory, but the full evaluation of the family is pending. The mother at first glance presents apparent hypertelorism.

This patient further confirms that the conditions associated with the deletion 22q11.2 not only includes most of the patients with DiGeorge, velocardiofacial, and conotruncal anomaly face syndromes, but also some patients with the Opitz GBBB phenotype as reported by the group in Philadelphia. This is in agreement with the recent publication of genetic heterogeneity in Opitz GBBB phenotype with one locus on Xp22, and a second locus on 22q11.2 [Robin et al., 1995].

REFERENCES

McDonald-McGinn DM, Driscoll DA, Bason L, Christensen K, Lynch D, Sullivan K, Canning D, Zavod W, Quinn N, Rome J, Paris Y, Weinberg P, Clark BJ, Emanuel BS, Zackai EH (1995): Autosomal dominant "Opitz" GBBB syndrome due to a 22q11.2 deletion. Am J Med Genet 59:103-113.

Robin NH, Feldman GJ, Aronson AL, Mitchell HF, Weksberg R, Leonard CO, Burton BK, Josephson KD, Laxova R, Aleck KA, Allanson JE, Guion-Almeida ML, Martin RA, Leichtman LG, Price RA, Opitz JM, Muenke M (1995): Opitz syndrome is genetically heterogeneous, with one locus on Xp22, and second locus on 22q11.2. Nature Genet 11:459-461.

Zackai EH, McDonald-McGinn DM, Driscoll DA, Emanuel BS, Christensen KM, Chien P, Mahboubi S, Hubbard AM, Weinberg P, Clark BJ (1995): Respiratory symptoms may be the first presenting sign of a 22q11.2 deletion: A study of vascular rings. Platform presentation, 16th David W. Smith Workshop on Malformations and Morphogenesis, Big Sky, MT.

Yves Lacassie
Marta I. Arriaza
Department of Pediatrics
Louisiana State University Medical Center
and Children's Hospital
New Orleans, Louisiana

Address reprint request to Yves Lacassie, M.D., Department of Pediatrics, LSU Medical Center, 1542 Tulane Avenue T8-1, NewOrleans, LA 70112.